Journal of Medical Genetics, 1978, **15**, 208–211

Brachydactyly, distal symphalangism, scoliosis, tall stature, and club feet: a new syndrome

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SUMMARY Five members of a kindred with brachydactyly and distal symphalangism, normal stature, pes cavus, and scoliosis were ascertained. The pedigree was consistent with autosomal dominant inheritance. The combination of clinical and radiological features is believed to be distinct from those previously reported in patients with brachydactyly/symphalangism.

The syndrome of brachydactyly and symphalangism has been reviewed in detail by Bell (1951), Temtamy and McKusick (1969), and Herrmann (1974). The findings in the hands and feet in our patients were consistent with type Al brachydactyly (Bell, 1951), with rudimentary or short middle phalanges. Normal stature in one kindred with type A brachydactyly has been reported by Nissen (1932), and scoliosis was reported in another kindred with normal stature and type A brachydactyly (Pippow, 1942). Vertebral involvement and severe pes cavus in association with brachydactyly/symphalangism similar to, but more extensive than, Bell type Al have been reported (Kirmisson, 1898).

The family reported indicates further heterogeneity in type Al brachydactyly. The presence of brachydactyly with club feet may be an important clinical clue to the subsequent development of scoliosis and allow early recognition and treatment of scoliosis in these patients.

Case reports

The pedigree is set out in Fig. 1.

CASE IV. 2: PROBAND

This young woman was born at term after a normal pregnancy and delivery. Birthweight was 3200 g and length 51 cm. Her fingers were short at birth and her feet showed varus deformity which was progressive and required manipulation and splinting during childhood. At 7 years, surgical correction of cavus and forefoot adduction deformity was performed.

Radiographs of the dorsal spine, performed because of back pain, at age 9 years, disclosed vertebral anomalies and a presumptive diagnosis of Scheur-

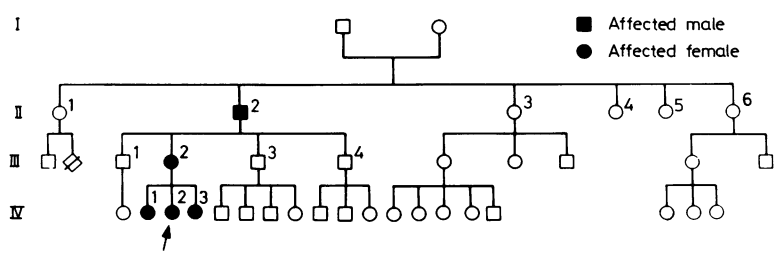

Fig. 1 *Family pedigree.*

Received for publication 4 May 1977

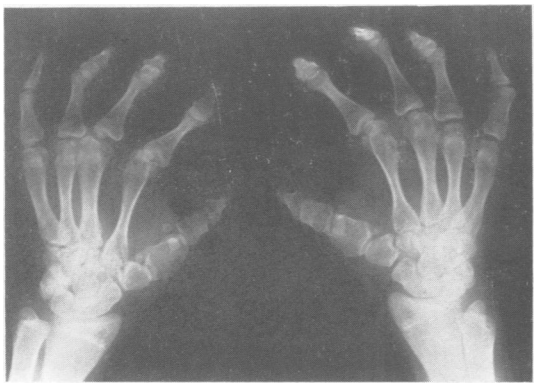

Fig. 2 *Proband aged 16 years: radiograph of hands.*

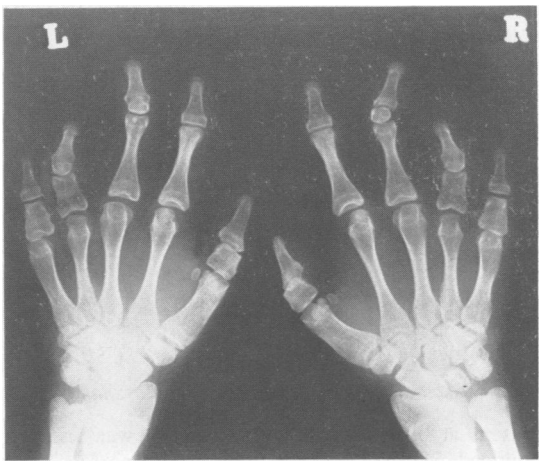

Fig. 3 *Elder sister of proband aged 17 years: hands.*

mann's disease was made, though she did not have kyphosis of the spine.

Scoliosis was diagnosed at 11 years. She presented at the Children's Hospital, Melbourne, when she was 16 years, for a Dwyer's operation for correction of scoliosis. Menarche occurred at 14½ years. Both her mother (height 176 cm) and her father (height 181 cm) were tall.

When examined, she was above the 98th centile for height, of slender build, with a moderate lumbar scoliosis convex to the left. Her face was oval, with small epicanthic folds. The fingers were short, with camptodactyly of the fifth finger. Thumbs were broad and thumb nails were short and broad though the nails on other fingers were normal. There was a single crease on each hand and single flexion creases on each finger. The forefeet were broad with pronounced cavus deformity.

Radiographs of the hands, at 16 years (Fig. 2), showed only 2 phalanges in the 2nd and 5th fingers. Proximal phalanges were of normal length, and distal phalanges were shaped like chess pawns. The middle phalanges of the 3rd and 4th fingers were rudimentary and square. In both hands the first metacarpals and the proximal phalanges of the thumb were short and square. The feet showed absent middle phalanges and pawn-shaped distal phalanges. The proximal phalanx of each hallux was square. Tarsal bones were larger than usual but otherwise normal. There was thoracolumbar scoliosis, and the vertebrae, particularly in the lumbar region, were large and square, and disc degenerative changes were pronounced for her age. The pelvis was ovoid and the iliac wings broad with horizontal acetabulae. Skull radiographs were normal.

CASE IV.1

The elder sister of the proband was 17 years when examined. She was born at term after a normal delivery, birthweight 3000 g and length 53 cm. Her early findings were similar to those of the proband. Cavus and varus deformities of both feet were present at birth. She had manipulation and splinting and three operations during childhood to correct these deformities. On examination, both feet were broad with pronounced cavus deformity. The toes were large and bulbous. Her hands showed pronounced brachydactyly, with camptodactyly of the fourth and fifth fingers and ulnar deviation of the distal phalanges of the middle fingers. The pattern of palmar creases was similar to that of the proband.

A thoracolumbar scoliosis became evident during her 14th year. Menarche occurred at 15 years. The scoliosis progressed during puberty and was of moderate severity at the time of examination. Her height was 174 cm, which is between the 25th and 50th centiles.

Radiographic findings were similar to the proband's. However, the proximal phalanges of ring and little fingers were short and square, similar to the first metacarpals of the proband (Fig. 3). There were only two phalanges in each finger. In the right hand only, a short square bone was interposed between proximal and distal phalanges of the middle finger. Radiographs of hands (Fig. 4) and pelvis at 3 months of age showed absent middle phalanges though there was a speck of calcification in the middle and ring finger of the left hand and middle finger of the right hand, consistent with an ossification centre for the middle phalanges. The pelvis had squared iliac wings with horizontal acetabulae.

CASE IV.3

The younger sister of the proband was 11 years when examined. Birthweight was 3700 g and length 56 cm. Changes in the hands similar to the proband's were

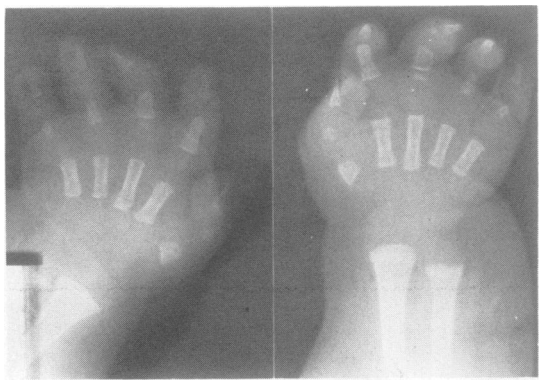

Fig. 4 *Elder sister of proband aged 3 months: hands.*

present at birth though foot deformity was less pronounced. She had a single operation to correct varus deformity at the age of 4 years. Recently she has had recurrent urinary tract infections but intravenous pyelogram and micturating cystourethrogram showed no abnormality.

On examination her height was between the 90th and 97th centiles. Scoliosis was absent. Her hands were similar to those of the proband's. Her feet showed mild cavus deformity only. Neurological examination was normal.

Radiographic findings were similar to those in the proband. However, the hands showed short rounded ossification centres interposed between proximal and distal phalanges (Fig. 5). In the thoracic spine the vertebrae were square and similar to the proband's. However, in the lumbar spine, the vertebrae were flattened.

CASE III.2

The mother of the proband was 48 years old when

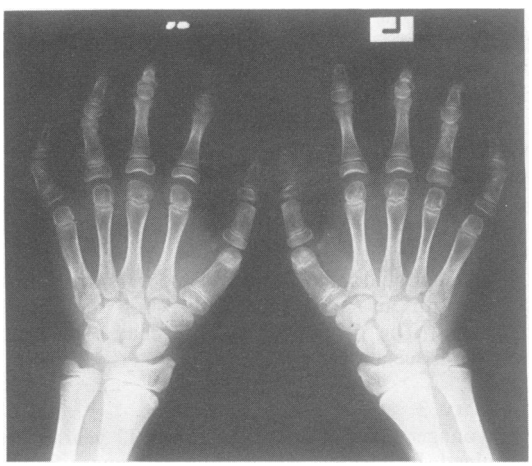

Fig. 5 *Younger sister of proband aged 11 years: hands.*

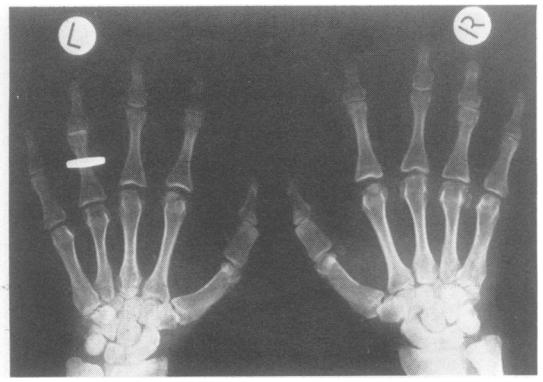

Fig. 6 *Mother of proband aged 48 years: hands.*

examined. Short fingers were present at birth though foot deformity was not present. Scoliosis had been noted at the age of 14 years but was not progressive.

She was 176 cm tall with mild thoracolumbar scoliosis. Her hands appeared identical to the proband's. Joint mobility was normal. The feet were broad with bulbous toes but were otherwise normal.

Radiographs of the hands showed absent middle phalanges, pawn-shaped distal phalanges, and relatively normal proximal phalanges except for that of the thumb which was short and rectangular (Fig. 6). Feet, thoracolumbar spine, pelvis, and skull were similar to the proband's.

CASE II.2

The proband's grandfather died at the age of 74 with cardiopulmonary disease which he had had for 6 years before his death. His daughter said that he was tall, height 188 cm, and had mild scoliosis. His fingers were short and his feet were broad but without pronounced cavus deformity. Radiographs were not available. He was the only one of his sibship ascertained with brachydactyly.

Genetics

This brachydactyly/symphalangism syndrome was expressed in 3 generations with 5 affected individuals. The pedigree (Fig. 1) was consistent with autosomal dominant inheritance. There was complete penetrance for brachydactyly but variable penetrance for pes cavus. Scoliosis was expressed at puberty. The younger sib had not yet reached puberty.

The first affected family member was the grandfather of the proband. His father was 36 years and mother 29 years at the time of his birth.

Discussion

Familial scoliosis has been reported as an isolated defect (Wynne-Davies, 1968) and associated with

heritable connective tissue disorders such as Marfan's syndrome. In this kindred, familial scoliosis occurred associated with tall normal stature, pes cavus, and brachydactyly similar to Bell type Al (Bell, 1951). The majority of reported cases of brachydactyly Al have been associated with short stature. However, normal stature and dominantly inherited brachydactyly were reported in one kindred with type A brachydactyly (Nissen, 1932). Scoliosis was reported in a further kindred with dominantly inherited brachydactyly and normal stature, though scoliosis occurred in other family members without brachydactyly (Pippow, 1942).

The findings in the reported family are similar to those reported for Kirmisson type brachydactyly/symphalangism where vertebral abnormalities and severe pes cavus are reported (Herrmann, 1974). The Kirmisson type syndrome differs in that finger nails and distal phalanges are absent.

In the family reported here, the onset of scoliosis has been recognised just before menarche. This pattern is similar to that of idiopathic scoliosis (Wynne-Davies, 1968). In the mother and older sister of the proband, progression of the scoliosis occurred during puberty but stabilised towards the end of the pubertal growth spurt. In the proband, the curve at presentation was such that operation was indicated to stabilise the scoliosis and prevent further progression into adult life.

The radiographs of the hands of the proband and her two sisters irregularly showed interposition of a centre of ossification between the proximal and distal phalanges of each hand. When these hand radiographs are compared with those in the atlas of skeletal maturation (Greulich and Pyle, 1959), for appropriate age, two different conclusions can be reached. The interposed bones may be large epiphyses of the distal phalanges. Alternatively, these bones may represent a composite of an epiphysis of a distal phalanx with an endochondral ossification centre for a vestigial middle phalanx. In this case, the most likely interpretation of the changes in the hands from IV.1 as a baby through IV.3 to the adult hand of III.2 is that they represent symphalangism of hypoplastic middle phalangeal ossification centres with distal phalanges. The striking feature of the syndrome is the chess pawn-shaped distal phalanges. From the series of radiographs it appears that these pawn-shaped phalanges arose from fusion of the interposed ossification centres with their distal phalanges.

The unusual shape of the vertebrae and the occurrence of scoliosis indicate a generalised connective tissue defect in this syndrome. Joint hyperlaxity was not a feature, suggesting that the defect was primarily expressed through the osseous system.

I am indebted to Dr K. Goard, Canberra, Australia and The Hospital for Sick Children, Great Ormond Street, London who provided radiographs of the family; to Dr Colin Jennings, Canberra, who allowed me to study his patients; and to Dr John Rogers, Professor D. M. Danks, Mr R. V. Dickens, Mr M. B. Menelaus, and Dr V. Mayne, Royal Children's Hospital, Melbourne for their help and encouragement with this study.

References

Bell, J. (1951). On brachydactyly and symphalangism. *Treasury of Human Inheritance,* **5** (1), 1–31.

Greulich, W. W., and Pyle, S. I. (1959). *Radiographic Atlas of Skeletal Development of the Hand and Wrist.* Stanford University Press, California.

Herrmann, J. (1974). Symphalangism and brachydactyly syndrome: report of the WL symphalangism-brachydactyly syndrome: review of literature and classification. *Birth Defects: Original Article Series,* **10,** No. 5, 23–53.

Kirmisson, E. (1898). Double pied bot varus, associeté des anklyoses congénitales des doigts et des osteils chez quartre membres d'une meme famille. *Revue d'Orthopédie et de Chirurgie de l'Appareil Moteur,* **9,** 392. (Cited by Herrmann, 1974.)

Nissen, K. I. (1932). A study in inherited brachydactyly. *Annals of Eugenics,* **5,** 281–301. (Cited by Bell, 1951.)

Pippow, G. (1942). Uber das Zusammentreffen von Wirbelgelenkaplasien und Brachydaktylie in einer Sippe. *Der Erbarzt,* **10,** 226–36. (Cited by Bell, 1951.)

Temtamy, S., and McKusick, V. A. (1969). Synopsis of hand malformations with particular emphasis on genetic factors. *Birth Defects: Original Article Series,* **5,** No. 3, 125–184.

Wynne-Davies, R. (1968). Familial idiopathic scoliosis. *Journal of Bone and Joint Surgery,* **50B,** 24–30.

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